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1: FGFR3 fibroblast growth factor receptor 3 (achondroplasia, thanatophoric dwarfism) [*Homo sapiens*]

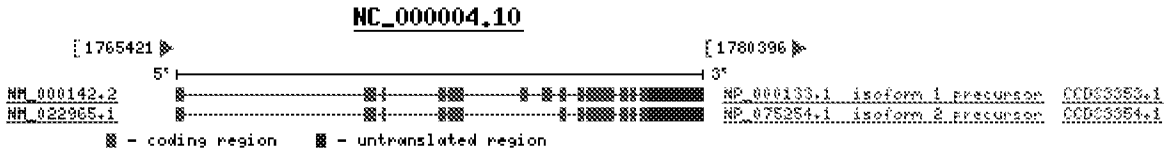
GeneID: 2261 updated 08-Jur

Summary

Official Symbol	FGFR3 provided by HG
Official Full Name	fibroblast growth factor receptor 3 (achondroplasia, thanatophoric dwarfism) provided by HG
Primary source	HGNC:3690
See related	Ensembl:ENSG00000068078; HPRD:00624; MIM:134934
Gene type	protein coding
RefSeq status	REVIEWED
Organism	*Homo sapiens*
Lineage	*Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammali Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Hor*
Also known as	ACH; CEK2; JTK4; CD333; HSFGFR3EX
Summary	The protein encoded by this gene is a member of the fibroblast growth factor receptor family, where amino acid sequence is highly conserved between members and throughout evolution. FGFR family members differ from one another in their ligand affinities and tissue distribution. A full-length representative protein would consist of an extracellular region, composed of three immunoglobulin-like domains, a single hydrophobic membrane-spanning segment and a cytoplasmic tyrosine kinase domain. The extracellular portion c the protein interacts with fibroblast growth factors, setting in motion a cascad of downstream signals, ultimately influencing mitogenesis and differentiation. This particular family member binds acidic and basic fibroblast growth hormon and plays a role in bone development and maintenance. Mutations in this gene lead to craniosynostosis and multiple types of skeletal dysplasia. Alternative splicing occurs and additional variants have been described, including those utilizing alternate exon 8 rather than 9, but their full-length nature has not be determined.

Genomic regions, transcripts, and products

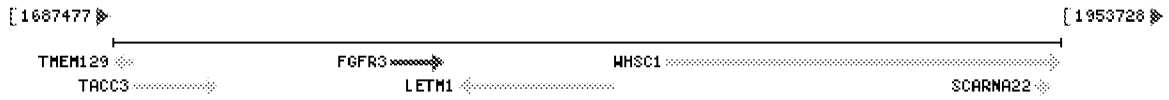
Go to reference sequence details Try our new Sequence V



Genomic context

chromosome: 4; Location: 4p16.3

See FGFR3 in MapV



Bibliography

Related Articles in PubMed

PubMed links

GeneRIFs: Gene References Into Function

1. The technique had a sensitivity and specificity of 100%. CONCLUSION: Hig resolution melting analysis is a simple, rapid, and sensitive one tube assay genotyping the FGFR3 gene.
2. No sequence variation was found, indicating that mutations in the "hot spot are not associated with nonsynostotic plagiocephaly.
3. Strong immunohistochemical expression of FGFR3, a superficial staining pa CK20, and a low proliferative activity define those papillary urothelial neopl low malignant potential that do not recur.
4. Observational study of genotype prevalence and gene-disease association. Navigator)
5. Observational study of gene-disease association. (HuGE Navigator)
6. Observational study of genetic testing. (HuGE Navigator)
7. FGFR3 mutation frequency was significantly associated with tumor grade.
8. over-expression of FGFR3 protein in many tumours compared to normal bla and ureteric controls. Increased expression was associated with mutation (mutant tumours)
9. MM Patients showing the t(4;14) chromosomal translocation at FGFR3 and genes had a significant elevation of serum crosslaps, reported to be the ma most reliably correlated with the extent of bone resorption
10. analysis of FGFR3 mutation in Muenke syndrome
11. a K650Q mutation in the FGFR3 gene may have a role in Acanthosis nigrica [case report]
12. Mutations were detected in 12 of 13 (92.3%) tumor tissues and 11 of 13 (8 urine samples from patients with superficial bladder cancer.
13. The group of low-grade noninvasive papillary tumours is defined by the pre

an FGFR3 mutation

14. FGFR3 mediates hematopoietic transformation by activating RSK2 in a two- fashion, promoting both the ERK-RSK2 interaction and subsequent phospho of RSK2 by ERK.

15. previously undescribed, heterozygous lysine to threonine mutation at codor the FGFR3 gene in familial acanthosis nigricans

16. Results suggest that a PLCgamma-STAT1 pathway mediates apoptotic signa FGFR3 in genetic dwarfism and chondrogenic cell lines.

17. the mutant S249C FGFR3 may have a role in bladder cancer

18. Mutations are a possible prognostic tool in survival of urothelial carcinoma upper urinary tract.

19. FGFR3 may represent a prognostic marker of chromosomally stable bladder with low malignant potential.

20. study showed that FGFR3 mutations appear to be common genetic alteratic multiple seborrheic keratoses with a varying interindividual mutation freque without specific intraindividual hot spots

21. the fibroblast growth factor family has a pleiotrophic function in human spermatogonia, which physiologically express FGFR3

22. Show preferential occurrence of FGFR3 mutations in seborrheic keratoses o head and neck. Increased age appears to be a risk factor for these mutatio

23. analysis of a Chinese family with autosomal dominant achondroplasia; disea locus was mapped to chromosome 4p16.3, where the FGFR3 gene is locate novel Ser217Cys mutation in exon 5 of FGFR3 that causes autosomal domi achondroplasia was identified

24. Although mutation K650M induces defective targeting of receptor, different mechanism characterized by receptor retention at plasma membrane, exce ubiquitylation and reduced degradation results from mutations of extracellu domain and stop codon.

25. Two patients with clinical and radiological findings of achondroplasia, who h most common FGFR3 missense mutations.

26. G380R mutation of this gene is common mutation associated with achondro

27. involvement of FGFR-3 in malignant hematopoiesis and FGFR-3 tyrosine kir CD34+ leukemic cells

28. data indicate that t(4;14)(p16;q32) and loss of fibroblast growth factor rec occurred at a very early stage of multiple myeloma and suggest that activa multiple myeloma SET domain protein may be transforming event of this translocation

29. Nucleotide 1138 in transmembrane domain of FGFR3 gene is the hot point mutation in ACH and hence its major pathologic cause.

30. FGFR3 expression is significantly associated with two important prognostic stage and grade. Tumours with FGFR3+/p53- phenotype seem to have a distinctive pathway in bladder tumorigenesis.
31. identified a heterozygous missense mutation that is predicted to cause a p. substitution in the tyrosine kinase domain and partial loss of FGFR3 functio
32. no evidence that mosaicism for mutations, normally associated with syndro forms of craniosynostosis, occur in single suture craniosynostosis
33. results indicate that FGFR 3 plays a crucial role in the accelerated proliferat MM carrying t(4;14)(p16.3;q32)
34. introduction of these mutated FGFR3s into ATDC5 cells downregulated PTHr expression and induced apoptosis with reduction of Bcl-2 expression
35. tumors with FGFR3 mutation appear to have distinctive clinical and biologic characteristics that may help in defining a population of patients for FGFR3 mutation screening
36. results give further support to the fact that the G380R mutation of FGFR-3 most common mutation causing achondroplasia in different populations
37. These results suggest that constitutive levels of both FGFR1 and FGFR3, bu FGFR4 are essential for FGF-stimulated anchorage-independent growth of S cells.
38. CHEK2 mutation has a role in hereditary breast cancer
39. Acanthosis nigricans with achondroplasia due to Gly380Arg mutation in FGF
40. Reciprocal relationship in gene expression between FGFR1 and FGFR3 in co tissue plays an important role in the progression of the carcinomas to malig
41. Mutations were detected in 23 of 27 (85%) adenoid seborrheic keratoses. F mutations were the most frequent mutation type.
42. Identification and characterization of an alternatively spliced isoform
43. Parathyroid hormone receptor type 1/Indian hedgehog expression is preser the growth plate of human fetuses affected with activating mutations in this protein
44. there is an FGFR3 mutation with a demonstrated deregulatory mechanism a alternative splicing in the absence of t(4;14) in multiple myeloma patients
45. Cherubism was mapped to region 4p16.3. Because of the associated craniosynostosis, we excluded the FGFR3 gene as a candidate gene for che
46. defective differentiation of chondrocytes is the main cause of longitudinal b growth retardation in FGFR3-related human chondrodysplasias
47. Differences in spatial patterns of FGFR expression in normal skin may gene functional diversity in response to FGFs, and in wounded skin FGFs may fur wound healing via induced FGFRs.

48. A missense mutation in FGFR3 resulted in skeltal dysplasia distinct from thanatophoric dysplasia.
49. FGFR3 is an important cell survival and antiapoptotic factor for multiple my cells
50. the G370C and S371C mutant receptors spontaneously dimerize in the corr spatial orientation required for effective signal transduction, whereas the 3 mutants, like the WT receptor, may achieve this orientation only on ligand
51. Inhibition of FGFR3 in myeloma cell lines was associated with morphologic, phenotypic, and functional changes typical of plasma cell differentiation, inc increase in light-chain secretion and expression of CD31, followed by apopt
52. FGFR3IIIS may regulate FGF and FGFR trafficking and function, possibly contributing to the development of a malignant phenotype
53. Over expression of FGFR3 is associated with urinary tract carcinoma progre
54. Gly380 --> Arg mutation does not alter dimerization energetics of FGFR3 transmembrane domain in detergent micelles or lipid bilayers. This indicate pathogenesis in achondroplasia cannot be explained simply by higher dimer of mutant FGFR3.
55. phosphorylation is essential for FGFR3 ubiquitylation, but is not sufficient to downregulation of its internalization resistant mutants
56. mutations in bladder cancer previously identified in non-lethal skeletal diso
57. presence of a Pro250Arg mutation predisposed to an increased transcranial reoperation rate...on the basis of raised intracranial pressure...in apparentl "isolated" coronal synostosis
58. Review. The role of FGFR3 in endochondral ossification and mutations leadi chondrodysplasia are reviewed.
59. First quantitative measurement of mutant receptor tyrosine kinase (RTK) stabilization in the membrane domain environment of FGFR3 shows the pro effect of resultant increase in the dimer fraction on RTK-mediated signal transduction.
60. Results suggest a novel interaction between the SOCS1 and SOCS3 protein the FGFR3 signaling pathway.
61. PRO-001 antibody is a potent and specific inhibitor of FGFR3 and deserves study for the treatment of FGFR3-expressing myeloma.
62. MIP-1 alpha promoter function, gene expression, and protein secretion wer down-regulated following inhibition of FGFR3 signaling.
63. PIK3CA mutations were strongly associated with FGFR3 mutations in superf papillary bladder tumors.
64. The R248C mutation appears to be a hot spot for FGFR3 mutations in epide nevi.

65. FGFR3 gene mutation is found in thanatophoric dysplasia type 1 and bilater renal dysplasia.
66. Data indicate that after endocytosis, fibroblast growth factor receptor (FGFI its bound ligand, FGF1, are sorted mainly to the recycling compartment, wh FGFR1-3 with ligand are sorted mainly to degradation in the lysosomes.
67. For the first time in humans, the expression of basic fibroblast growth factc (bFGF) and its receptors FGFR-2, FGFR-3, and FGFR-4 has been documente ovaries of second- and third-trimester fetuses.
68. Activating mutations of FGFR3 are associated with benign skin tumors.
69. Mutation in the FGFR3 is associated with progression of oral squamous cell
70. Patients with TWIST gene mutations may have more ophthalmic abnormali including more strabismus, ptosis, NLDO, astigmatism, vertical deviations, amblyopia compared with patients with FGFR3 gene mutations.
71. The FGFR3-associated coronal synostosis syndrome (Muenke craniosynosto caused by a point mutation (C749G) on the FGFR3 gene resulting in a Pro2 substitution.
72. IGF-1 prevents the apoptosis induced by FGFR3 mutation through the PI3K pathway and MAPK pathway
73. FGFR3 mutation status and loss of 9q is associated with early-stage bladde carcinomas
74. The present studies show that MUC1 associates with FGFR3.
75. double mutation in FGFR3 encoding GLY380LYS is responsible for Hypochondroplasia.
76. findings indicate that: (1) FGFR3 mutations occur in mosaicism and can cau epidermal nevi and (2) other genes are involved in epidermal nevi
77. The cell model will be useful for the study of FGFR3 function in cartilage stu and future therapeutic approaches in chondrodysplasias.
78. distribution in normal endocrine cells and related tumors of the gastroenteropancreatic system; immunoreactive in duodenal G cells
79. Mutations in growth factor receptor 3 is associated with the pathogenesis o urothelial cell carcinoma
80. fibroblast growth factor receptor 3 has a role in trafficking and signaling
81. Over-expression of FGFR3 may play an important role in liver carcinogenes
82. fibroblast growth factor receptor 3 mutations have a role in development of bladder cancer
83. FGFR3 mutations do not seem to play a role in bladder cancer progression
84. We identified a novel ETV6 partner gene, fibroblast growth factor receptor 3 (FGFR3), in a patient with peripheral T-cell lymphoma (PTCL) with a t(4;12

(p16;p13) translocation.

85. FGFR3 and Tp53 mutations do not appear to be associated with progressior T1G3 transitional bladder carcinomas

86. A child who has hypochondroplasia due to an N540K mutation and who has temporal lobe dysgenesis.

87. the importance of the immature FGFR3 proteins as mediators of an abnorm signaling in thanatophoric dysplasia type II

88. FGFR3 mutations were associated with low-stage, low-grade urothelial carc of the blader.

89. strong correlation beween mutations of FGFR3 and disturbances of skeletal REVIEW

90. fibroblast growth factor receptor 3 activation is regulated by cytoplasmic ty kinase Pyk2

91. The detection of FGFR3 mutations in FUH (Flat Urothelial Hyperpalsias) sup the role of this lesion as precursor of papillary bladder cancer.

92. These results suggest that intracellular domain mutations define a distinct r by which mutated FGFR3 could disrupt bone development.

93. alternative splicing of FGFR3IIIb results in a secreted isoform that inhibits F induced proliferation

94. interacts with adapter protein SH2-B, and has a role in STAT5 activation

Interactions

Description .....

Product	Interactant	Other Gene	Complex	Source	Pubs
NP_000133.1	NP_067007.3	C6orf47		HPRD	Pu
NP_000133.1	Fibroblast growth factor 1	FGF1		HPRD	Pu
NP_000133.1	Fibroblast growth factor 8	FGF8		HPRD	Pu
NP_000133.1	Fibroblast growth factor 9	FGF9		HPRD	Pu
NP_000133.1	NP_002077.1	GRB2		HPRD	Pu

General gene information

Markers

WI-15208(e-PCR)
Links: UniSTS:34762
Alternate names: EST318764; RH59111

G15851(e-PCR)
Links: UniSTS:43116
Alternate names: CHLC.UTR_02040_M64347; CHLC.UTR_02040_M64347.P56111

RH18137(e-PCR)
Links: UniSTS:91412

GDB:187013(e-PCR)
Links: UniSTS:155533

GDB:454672(e-PCR)
Links: UniSTS:157443

GDB:454675(e-PCR)
Links: UniSTS:157444

GDB:581559(e-PCR)
Links: UniSTS:157858

GDB:585477(e-PCR)
Links: UniSTS:157890

GDB:681581(e-PCR)
Links: UniSTS:158627

Genotypes

See FGFR3 SNP GeneView Report
See FGFR3 SNP Genotype Report

Phenotypes

Achondroplasia
MIM: 100800
Bladder cancer
MIM: 109800
CATSHL syndrome
MIM: 610474
Cervical cancer, somatic
MIM: 603956
Colorectal cancer, somatic
MIM: 109800
Crouzon syndrome with acanthosis nigricans
MIM: 134934
Hypochondroplasia
MIM: 146000
LADD syndrome
MIM: 149730
Muenke syndrome
MIM: 602849
Nevus, keratinocytic, nonepidermolytic
MIM: 162900
Thanatophoric dysplasia, types I and II
MIM: 187600

Pathways

KEGG pathway: MAPK signaling pathway
04010
KEGG pathway: Regulation of actin cytoskeleton
04810
Reactome Event: Signaling by FGFR
190236

Homology

Mouse, Rat
Map Viewer

GeneOntology

Provided by

Function	Evidence
ATP binding	IEA
fibroblast growth factor receptor activity PubMed 7923141	NAS
identical protein binding	

PubMed 14732692	IPI
nucleotide binding	IEA
protein tyrosine kinase activity	IEA
receptor activity	IEA
transferase activity	IEA

Process	Evidence
JAK-STAT cascade PubMed 10918587	TAS
MAPKKK cascade PubMed 10918587	TAS
cell growth	NAS
fibroblast growth factor receptor signaling pathway PubMed 10918587	TAS
protein amino acid phosphorylation	IEA
sensory perception of sound	IEA
skeletal development PubMed 8601314	TAS

Component	Evidence
integral to membrane	IEA
integral to plasma membrane PubMed 10918587	TAS
plasma membrane PubMed 11294897,16597617	EXP

General protein information

Preferred Names
- fibroblast growth factor receptor 3

Names
- fibroblast growth factor receptor 3
- tyrosine kinase JTK4
- hydroxyaryl-protein kinase

NP_000133.1
EC 2.7.10.1
NP_075254.1
EC 2.7.10.1

NCBI Reference Sequences (RefSeq)

RefSeqs maintained independently of Annotated Genomes

These reference sequences exist independently of genome builds.

mRNA and Protein(s)

1. NM_000142.3→NP_000133.1 fibroblast growth factor receptor 3 isoform 1 precursor

Description	Transcript Variant: This variant (1) is missing alternatively spliced exon 8 but utilizes alternatively spliced exon 9, resulting in isoform (1) with the IIIc-type C-terminal half of the IgIII domain.
Source sequence(s)	AB209441,AC016773,BC153824
Consensus CDS	CCDS3353.1
UniProtKB/Swiss-Prot	P22607

Conserved Domains (5) summary

cd00931 Location:151–243 Blast Score:204	IGcam; Immunoglobulin domain cell adhesion molecule (cam) subfamily; members are components of neural cell adhesion molecules (N-CAM L1), Fasciclin II and the insect immune protein Hemolin. The subfamily also includes receptor domains such as as the...
cd05100 Location:459–753 Blast Score:1597	PTKc_FGFR3; PTKc_FGFR3: Protein Tyrosine Kinase (PTK) family; Fibroblast Growth Factor Receptor 3 (FGFR3); catalytic (c) domain. The PTKc family is part of a larger superfamily that includes the catalytic domains of other kinases such as protein serine/threonine...
pfam07714 Location:472–748 Blast Score:987	Pkinase_Tyr; Protein tyrosine kinase.
cl00093 Location:253–346 Blast Score:132 Location:53–110 Blast Score:103	IG; Immunoglobulin domain family; members are components of immunoglobulins, neuroglia, cell surface glycoproteins, such as, T-cell receptors, CD2, CD4, CD8, and membrane glycoproteins, such as, butyrophilin and chondroitin sulfate proteoglycan core...

2. NM_022965.2→NP_075254.1 fibroblast growth factor receptor 3 isoform 2 precursor

Description	Transcript Variant: This variant (2) does not contain alternatively spliced exons 8 or 9, resulting in a loss of the C-terminal half of the IgIII domain. In addition, this variant is missing alternatively spliced exon 10 which encodes the transmembrane region, suggesting a soluble receptor.
Source sequence(s)	AB209441,AC016773,AF245114
Consensus CDS	CCDS3354.1

Conserved Domains (4) summary

cd00931 Location:151–243 Blast Score:203	IGcam; Immunoglobulin domain cell adhesion molecule (cam) subfamily; members are components of neural cell adhesion molecules (N-CAM L1), Fasciclin II and the insect immune protein Hemolin. The subfamily also includes receptor domains such as as the...
pfam07714 Location:360–636 Blast Score:964	Pkinase_Tyr; Protein tyrosine kinase.
cl00093 Location:53–110	IG; Immunoglobulin domain family; members are components of immunoglobulins, neuroglia, cell

RefSeqs of Annotated Genomes: Build 36.3

The following sections contain reference sequences that belong to a specific genome build.

Reference assembly

Genomic

1. NC_000004.10 Reference assembly
   - Range: 1765421..1780396
   - Download: GenBank FASTA Sequence Viewer (beta)
2. NT_006081.18
   - Range: 301275..316250
   - Download: GenBank FASTA Sequence Viewer (beta)

Alternate assembly (based on Celera assembly)

Genomic

1. AC_000047.1 Alternate assembly (based on Celera assembly)
   - Range: 1708138..1723112
   - Download: GenBank FASTA Sequence Viewer (beta)
2. NW_921918.1
   - Range: 1708138..1723112
   - Download: GenBank FASTA Sequence Viewer (beta)

Alternate assembly (based on HuRef)

Genomic

1. AC_000136.1 Alternate assembly (based on HuRef)
   - Range: 1753471..1738498, complement
   - Download: GenBank FASTA Sequence Viewer (beta)
2. NW_001838896.2
   - Range: 2015637..2000664, complement
   - Download: GenBank FASTA Sequence Viewer (beta)

Related Sequences

Nucleotide	Protein

Genomic	AC016773.8 (170070..185045)	None
Genomic	AF487554.1	AAM22078.1
		AAM22079.1
Genomic	AY768549.1	AAU89726.1
Genomic	CH471131.2	EAW82562.1
		EAW82563.1
		EAW82564.1
		EAW82565.1
		EAW82566.1
		EAW82567.1
		EAW82568.1
Genomic	S76733.1	AAB33323.1
Genomic	U22410.1	AAA67781.1
mRNA	AB209441.1	BAD92678.1
mRNA	AF238374.1	AAF97749.1
mRNA	AF245114.1	AAF63380.1
mRNA	AF369211.1	AAK54727.1
mRNA	AF369212.1	AAK54728.1
mRNA	AF369213.1	AAK54729.1
mRNA	BC121175.2	AAI21176.1
mRNA	BC128610.1	AAI28611.1
mRNA	BC153824.1	AAI53825.1
mRNA	M58051.1	AAA52450.1
mRNA	M59374.1	AAA63209.1
mRNA	M64347.1	AAA58470.1
mRNA	X84939.1	CAA59334.1
Synthetic	BC166684.1	AAI66684.1

Protein Accession	Links	
P22607.1	GenPept	UniProtKB/Swiss-Prot
Q0IJ44	GenPept	UniProtKB/TrEMBL
Q59FL9	GenPept	UniProtKB/TrEMBL
Q8NI15	GenPept	UniProtKB/TrEMBL
Q8NI16	GenPept	UniProtKB/TrEMBL
Q96T34	GenPept	UniProtKB/TrEMBL
Q96T35	GenPept	UniProtKB/TrEMBL
Q96T36	GenPept	UniProtKB/TrEMBL
Q9NRB6	GenPept	UniProtKB/TrEMBL

Additional Links

- MIM 134934
- GeneTests for MIM: 100800
- GeneTests for MIM: 134934
- HPRD 00624
- UniGene Hs.1420

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